Movement Disorders
CLINICAL PRACTICE
CLINICAL VIGNETTE

Gelastic Cataplexy in Niemann Pick Type C

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Cataplexy delineates a brief episode of generalized loss of muscle tone without alteration in consciousness. When cataplexy is triggered by laughter, the term "gelastic" is utilized. Gelastic cataplexy and vertical supranuclear gaze palsy (VSGP) are classical features of Niemann Pick type C (NPC) disease.[1]

Herein, we report on a case of NPC with typical episodes of gelastic cataplexy with video documentation.

Case

A 14-year-old boy presented to us with movement disorder, mental decline, and gait disturbance. He was well and had a normal birth and development up to age 10, when deterioration in cognitive function and gait impairment began. He also developed abnormal postures in the neck and limbs. During the last year, he had episodes of generalized atonia without loss of consciousness, triggered by laughing (gelastic cataplexy; Video 1). His parents were first cousins. Physical examination revealed splenomegaly. On neurological examination, he was alert and could follow simple commands. Speech was unintelligible because of severe dysarthria. He had a VSGP (Video 2) and mild dystonic posture in the neck and extremities with limb and gait ataxia (Videos 3 and 4).

Routine blood tests, EEG, and brain MRI were normal. Genetic testing showed a homozygous mutation in the *NPC1* gene (g.21119403T>C NM_000271.4: c.2827A>G p.(Ile943Val) Exon 19).

Discussion

NPC is a rare, autosomal-recessive lysosomal storage disorder with a prevalence of 1:100,000 live births. It is caused by mutations in either the *NPC1* (in approximately 95% of cases) or the *NPC2* gene.[2] Its clinical features encompass a combination of visceral, neurological (movement disorders, cerebellar ataxia, seizure, and cognitive impairment), and psychiatric signs and symptoms. Clinical presentation of NPC is impressed by age at disease onset, with more common cortical signs (cognitive impairment and psychosis) in adult-onset cases.

Gelastic cataplexy and VSGP are specific signs of NPC.[1,2] Gelastic cataplexy is observed in almost 50% of all patients with NPC and may develop during the course of the disease or rarely be the presenting symptom.[3]

Cataplexy reflects a rapid eye movement (REM)-sleep-related phenomenon involving multiple neurotransmitter systems which regulate sleep and postural muscle tone. It is commonly a component of the narcoleptic syndrome alongside daytime sleepiness, sleep paralysis, and vivid hallucinations at the beginning and end of sleep.[4]

Cataplexy is thought to represent brief episodes of REM sleep paralysis during wakefulness and probably involves the same neural networks that generate normal muscle atonia in REM sleep. Treatment of cataplexy by tricyclic antidepressants is often successful.[3] This most likely reflects the general suppression of REM sleep by this class of medications attributed to activation of monoaminergic pathways.

Additionally, hypocretin excites motor neurons and helps to maintain muscle tone by activating the monoaminergic pathway. In NPC, the accumulation of lipid-storage products in the nervous system might affect the hypocretin-producing cells in the hypothalamus, reducing its level.[5]

In conclusion, this case demonstrates the importance of gelastic cataplexy as an important clue in diagnosis of NPC especially when accompanied by VSGP.

Acknowledgments

The authors acknowledge the patient and his family members for consenting to participate in this study.

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(1) Research Project: A. Conception, B. Organization, C. Execution; (2) Statistical Analysis: A. Design, B. Execution,

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Keywords: cataplexy, gelastic cataplexy, Niemann Pick type C, supranuclear vertical gaze palsy.
Relevant disclosures and conflicts of interest are listed at the end of this article.
Received 21 February 2019; revised 4 May 2019; accepted 12 May 2019.
Published online 26 June 2019 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/mdc3.12786

C. Review and Critique; (3) Manuscript: A. Writing of the First Draft, B. Review and Critique.

N.Y.: 1A, 1C, 3B

A.F.: 3B

A.E.L.: 3B

M.R.: 1A, 1B, 3A

Disclosures

Ethical Compliance Statement: The authors confirm that the approval of an institutional review board was not required for this work. The authors obtained written consent from the patient's father. We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this work is consistent with those guidelines.

Funding Sources and Conflicts of Interest: The authors report no sources of funding and no conflicts of interest.

Financial Disclosures for previous 12 months: A.F. receives consultancy fees from AbbVie, Medtronic, Boston Scientific, Sunovion, Chiesi farmaceutici, UCB, and Ipsen; receives honoraria from AbbVie, Medtronic, Boston Scientific, Sunovion, Chiesi farmaceutici, UCB, and Ipsen; receives grant support from University of Toronto, Weston Foundation, AbbVie, Medtronic, and Boston Scientific; and sits on advisory boards for AbbVie, Boston Scientific, and Ipsen.

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Supporting Information

Supporting information may be found in the online version of this article.

Video S1. This video shows a typical episode of gelastic cataplexy with head and neck drop triggered by laughing.

Video S2. The video demonstrates the vertical gaze palsy overcome by oculocephalic maneuver.

Video S3. Finger dysmetria on finger-to-nose test and mild dystonic posture of upper limbs are shown in this video.

Video S4. This video shows the patient's gait, which is dystonic and ataxic; he is unable to walk in tandem.